

# *BRCA1* and *BRCA2* mutation screening in the Breast CFR

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for the BreastCFR

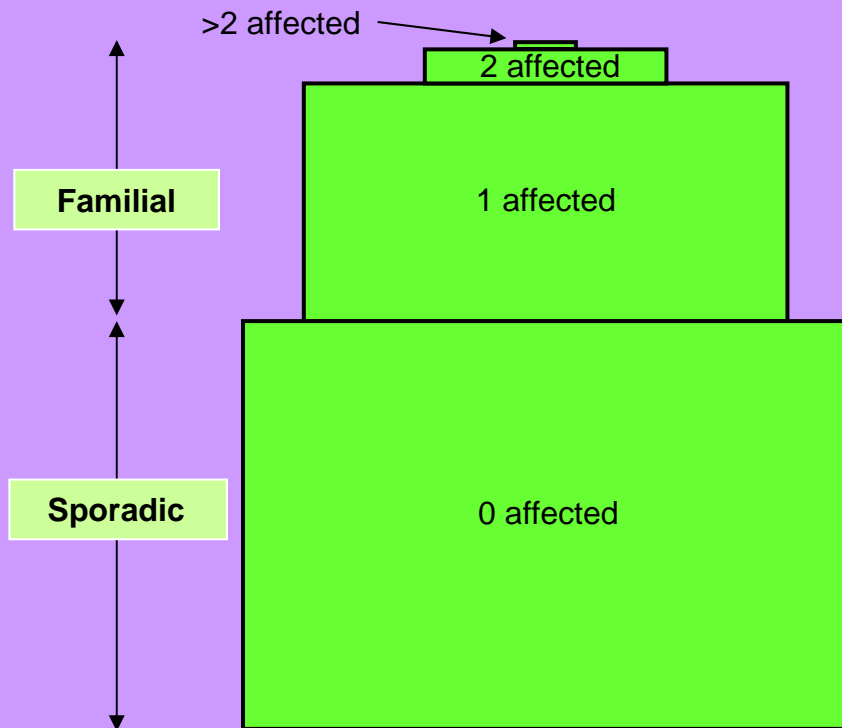
## ***BRCA1* and *BRCA2* mutation screening in the Breast CFR**

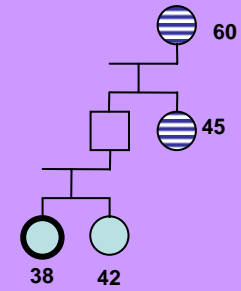
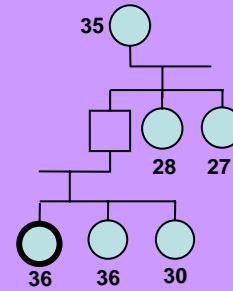
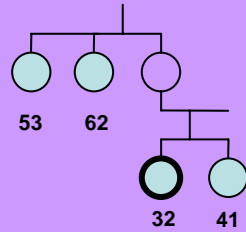
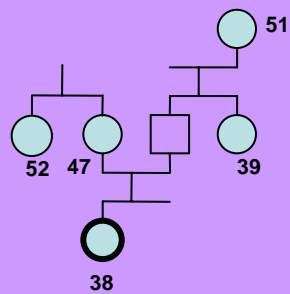
	<b>Total</b>
<b>Tested for BRCA1</b>	9360
Affected carrier	549
Unaffected carrier	439
Total carriers	<b>981</b>
<b>Tested for BRCA2</b>	7736
Affected carrier	307
Unaffected carrier	243
Total carriers	<b>550</b>

Funded via

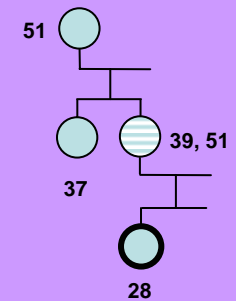
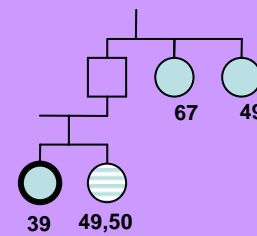
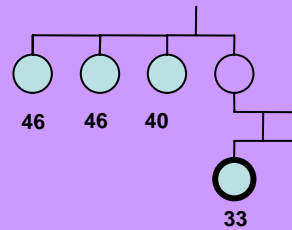
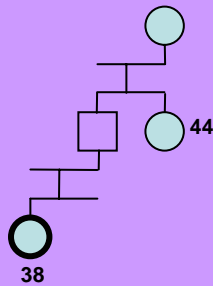
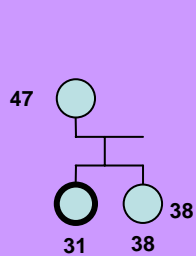
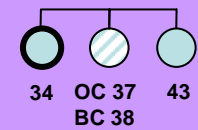
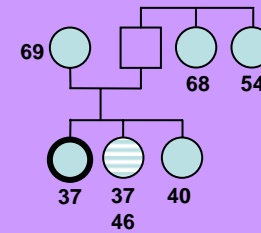
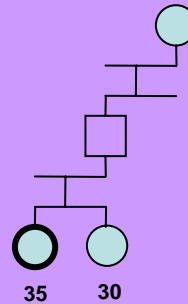
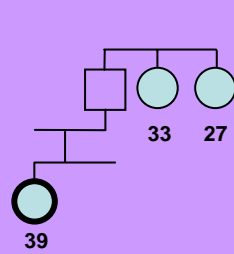
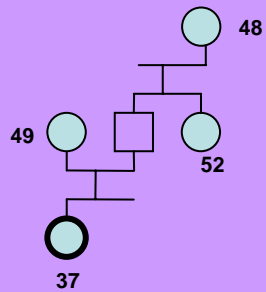
- Local funding
- AJ supplement
- CDC/NIH supplement
- RO1s

## ***BRCA1* mutation screening in cases**

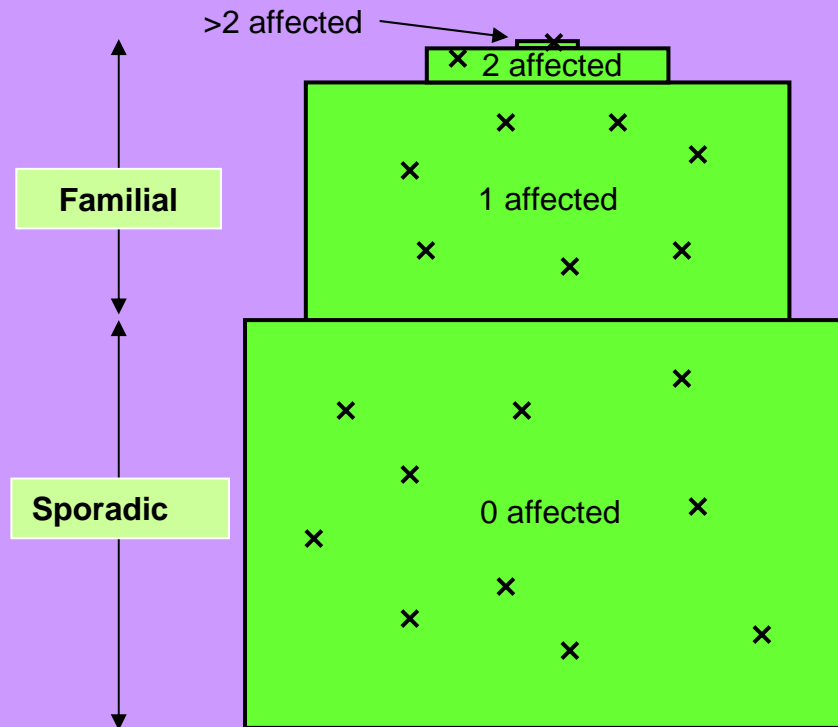




## ***BRCA1* and *BRCA2* mutation screening in the Breast CFR**

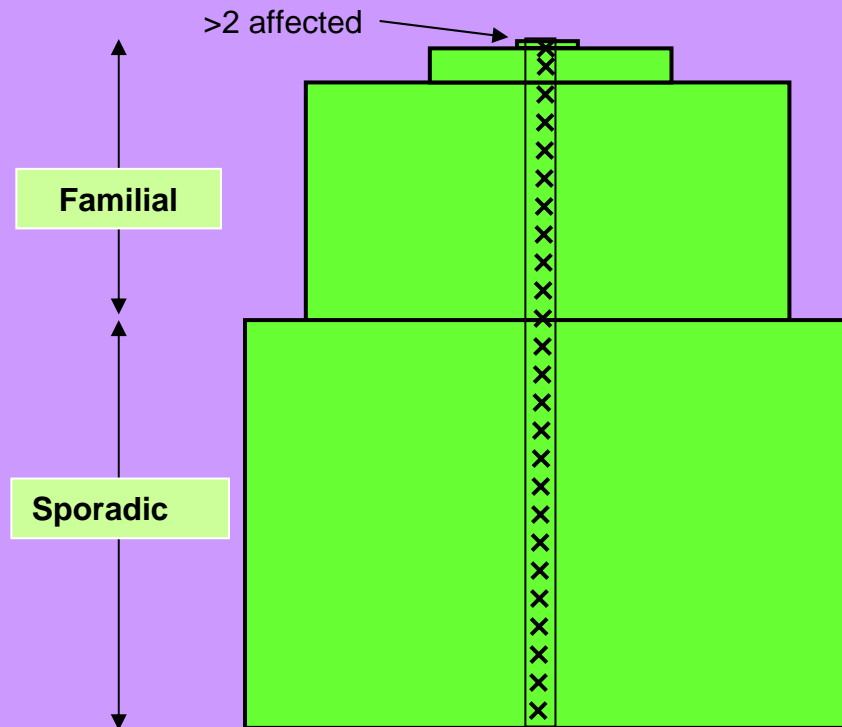


## ***BRCA1* mutation screening in cases**



Majority of *BRCA1* mutation carriers do not have a family of breast cancer.

## ***BRCA1* mutation screening in cases**



Expanded screening in multiple-case families with early onset cancer.

## Non-familial causes

Breast cancer is a “common” disease – how many of these multiple case families could have been caused by factors that are not correlated in family members ?

Using simulation based on the age of the relatives and the age specific incidence rates for the population, then ....

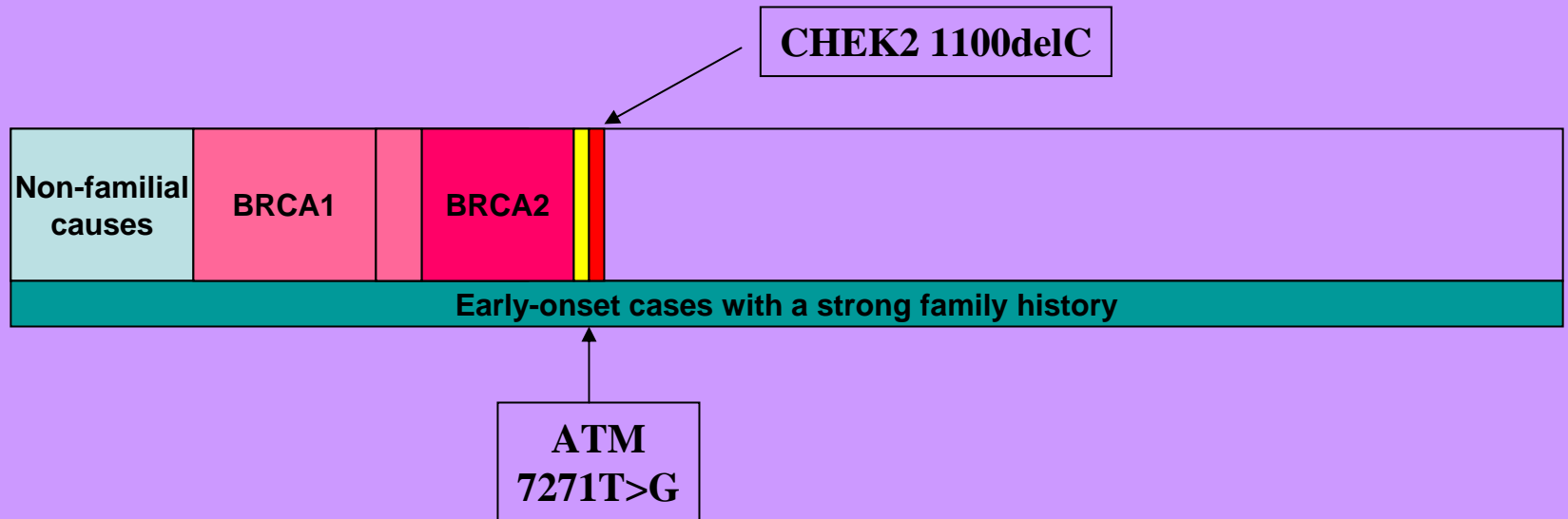
...even if there were no familial causes of early-onset breast cancer, 11% of cases of early onset breast cancer would have had a strong family history.

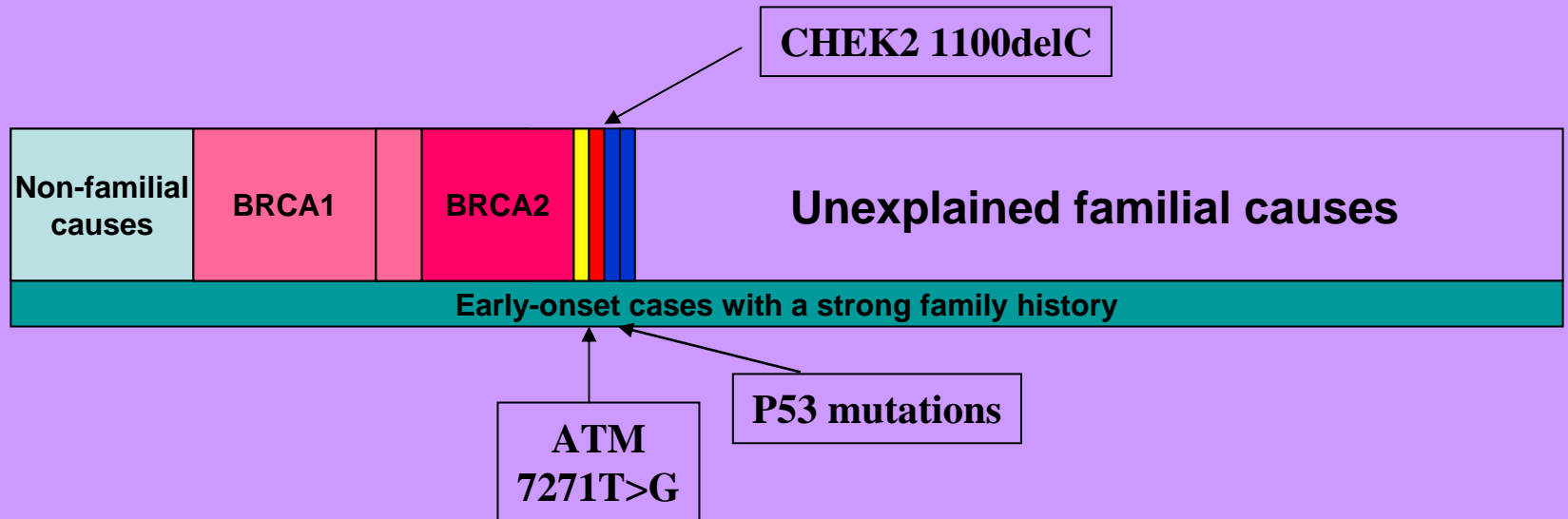
Non familial causes	Due to familial causes.
Early-onset cases with a strong family history	

# What Causes Early-Onset Breast Cancer Associated with a Strong Family History?

Non familial causes	BRCA1		BRCA2	
Early-onset cases with a strong family history				

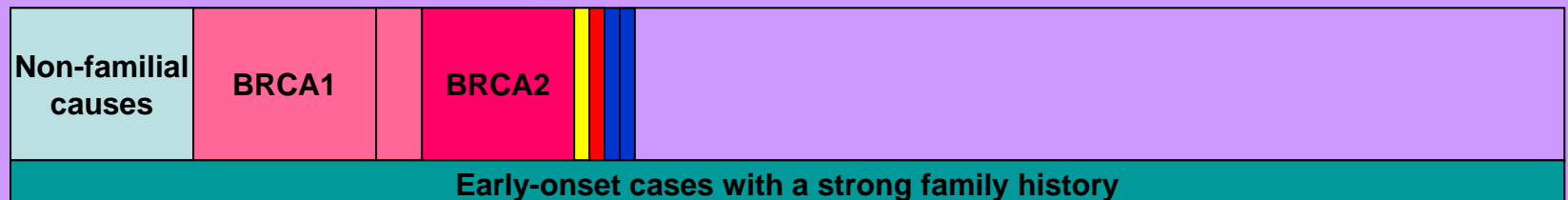






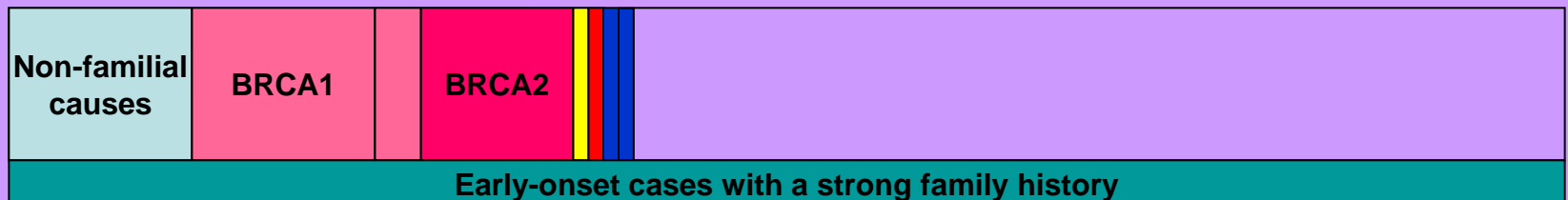
The majority of multiple-case families with early-onset breast cancer are **NOT** due to *BRCA1* and *BRCA2* mutations.

...or we do not know how to look for them all yet



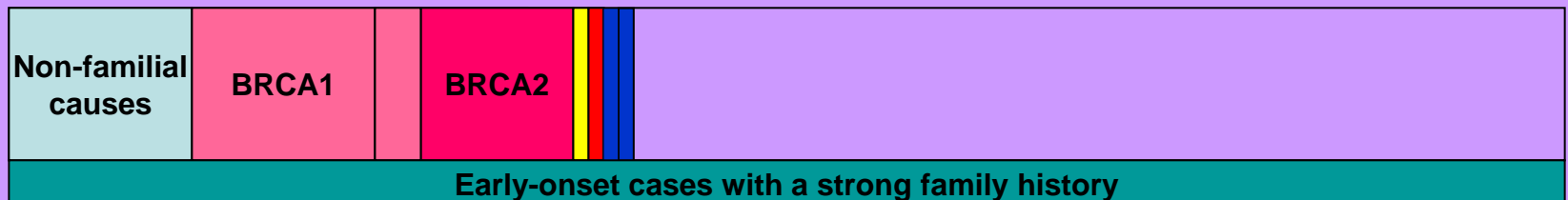
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...expanded screening in other selected groups

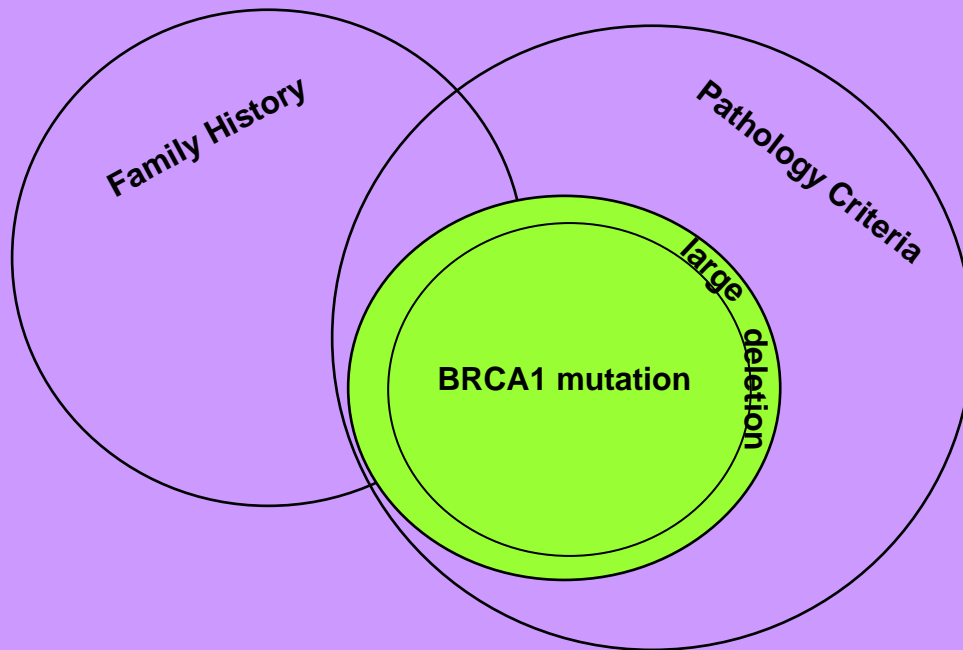


The majority of multiple-case families with early-onset breast cancer are **NOT** due to *BRCA1* and *BRCA2* mutations.

...tumour morphology



## Probands diagnosed under the age of 40 years



mitotic index greater than 50/10HPF

nuclear pleomorphism = 3

tubule formation = 3

syncytial growth pattern = 1 (present)

pushing margin = 1 (present)

circumscribed = 1 (present)

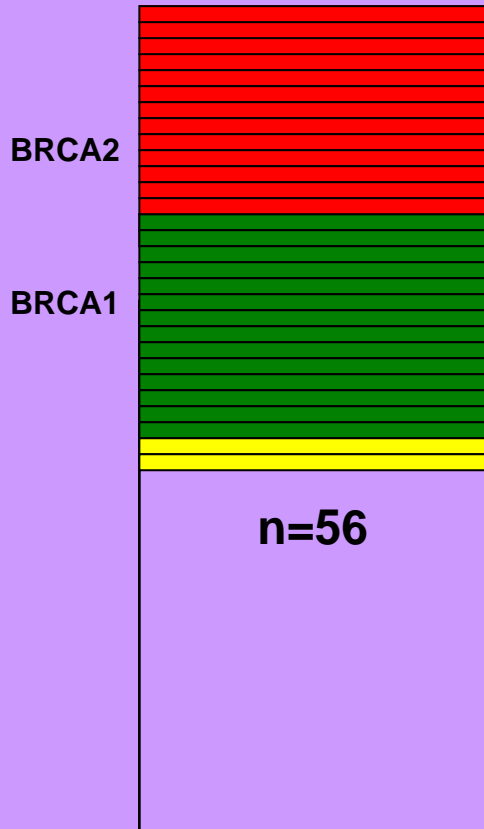
lymphocytic infiltrate (II) = 1 or 2 (moderate OR intense)

growth pattern (primary or secondary) = 3 (trabecular)

necrosis = 1 (present)

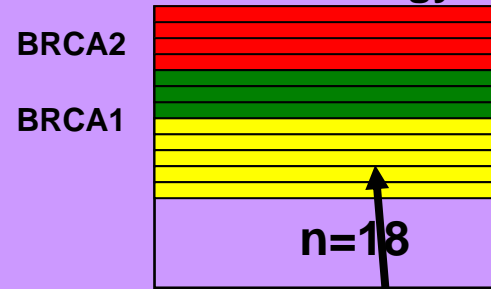
# Nth California Breast CFR

2+ family history  
(first degree relatives)



del1A-exon 24  
del1A-exon 17

5+ Pathology



del exon 17  
del1A-exon 17  
del exon 13  
dup exon 2-9  
dup exon 10

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